Analysis of protein-coding mutations in hiPSCs and their possible role during somatic cell reprogramming.

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Authors: Sergio Ruiz, Athurva Gore, Zhe Li, Athanasia D Panopoulos, Nuria Montserrat, Ho-Lim Fung, Alessandra Giorgetti, Josipa Bilic, Erika M Batchelder, Holm Zaehres, Hans R Scholer, Kun Zhang, Juan Carlos Izpisua Belmonte

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Public Summary:
In this work we characterized 17 genes that found mutated in human iPS cells, and showed that the majority of these mutations did not affect the efficiency of reprogramming.

Scientific Abstract:
Recent studies indicate that human-induced pluripotent stem cells contain genomic structural variations and point mutations in coding regions. However, these studies have focused on fibroblast-derived human induced pluripotent stem cells, and it is currently unknown whether the use of alternative somatic cell sources with varying reprogramming efficiencies would result in different levels of genetic alterations. Here we characterize the genomic integrity of eight human induced pluripotent stem cell lines derived from five different non-fibroblast somatic cell types. We show that protein-coding mutations are a general feature of the human induced pluripotent stem cell state and are independent of somatic cell source. Furthermore, we analyse a total of 17 point mutations found in human induced pluripotent stem cells and demonstrate that they do not generally facilitate the acquisition of pluripotency and thus are not likely to provide a selective advantage for reprogramming.